Title 48
HEALTH AND HOSPITALS
Part V. Public Health Services
Subpart 19. Genetic Diseases Services

Chapter 63. Neonatal Screening

§6301. Eligibility

A. Any child born in or residing in the state of Louisiana shall be eligible for neonatal screening.

AUTHORITY NOTE: Promulgated in accordance with R.S. 40:1299.

HISTORICAL NOTE: Promulgated by the Department of Health and Human Resources, Office of Preventive and Public Health Services, LR 13:246 (April 1987).

§6303. Purpose, Scope, Methodology

A. Purpose and Scope. R.S. 40:1299.1, 2,and 3 require physicians to test Louisiana newborns for phenylketonuria, congenital hypothyroidism, sickle cell disease, biotinidase deficiency and galactosemia. The Office of Public Health (OPH) maintains a laboratory for performing screening tests for hyperphenylalanemia manifest in phenylketonuria (PKU), for thyroxine (T_4) and thyroid stimulating hormone (TSH) used in congenital hypothyroidism detection, hemoglobin identification for sickle cell disease and enzyme assays for detection of biotinidase deficiency and galactosemia. Definitive diagnostic tests are provided if the screening test is positive. The newborn screening battery may also be available through other approved laboratories (see Subsection G).

B. Methodology

1. Filter Paper Specimen Form, (Lab-10) used in blood specimen collection for neonatal screening, can be obtained at parish health units. There are two different types of Lab-10 forms which are color coded.

a. For patients covered by Medicaid, including those in the Kid-Med Program, blue border Lab-10 forms are used. There is no charge to private providers for these blue border forms. The Patient's Medicaid number (or Mother's number, if the patient has not been issued one) must be incidated on the form.

b. For private and non-Medicaid patients, red border Lab-10 forms are used. These red border Lab-10 forms are $18 each.

2. Private providers should order a mix of red and blue Lab-10 forms from their local parish health unit (or OPH Regional Office for certain areas) to match the Medicaid/Non-Medicaid composition of newborns to be screened at their facility. The Lab-10 forms must be completely filled out.

3. For non-Medicaid patients with a financial status of greater than 100 percent of the Poverty Guidelines as established by the Department of Health and Hospitals (DHH) and who attend a parish health unit for just the newborn screening service, the parent or guardian will be charged $18 upon registering at the parish health unit.

4. To ensure that specimens for testing are received within two to three days by the laboratory approved by OPH to perform newborn screening pursuant to the pertaining requirements of this Chapter, all such laboratories must provide mailing envelopes to submitting hospitals which guarantee a delivery time no longer than three days from mailing. An example of an acceptable minimum option would be the use of the United States Postal Service's Flat Rate Priority Mailing Envelopes. The use of all other companies and courier services providing this service are acceptable.

C. Policy for Predischarge and Repeat Screening

1. All hospitals that have maternity units shall institute and maintain a policy of screening al neonates before discharge regardless of their length of stay in the hospital. Newborns remaining in the hospital for an extended period should be screened initially no later than seven days after birth. Neonatal screening results on specimens collected from babies younger than 48 hours of age may not be valid because of a large percentage of false negative results particularly at 24 hours or less (16 percent)[1]. These newborns must be re-screened at the first medical visit, preferable between one and two weeks of age, but no later than the third week of life. Selection of 48 hours, though somewhat inconsistent with the American Academy of Pediatrics (AAP) Guidelines for Perinatal Care, Second Edition, was recommended by our Genetics Advisory Committee and the pediatric endocrinology consultants.

2. A newborn initially screened before 48 hours of age must receive a repeat screening no later than the third week of life. Repeat screening should be arranged by the primary pediatrician; however, it may be done by any primary healthcare provider or clinical facility qualified to perform newborn screening specimen collection. The blood specimen collection should be performed at the infant's first medical visit, regardless of age if they are seen at 48 hours of age or older. To ensure that neonates who need rescreening (due to initial unsatisfactory specimen, an initial collection performed on a baby less than 48 hours old) actually receive the repeat test, hospitals with maternity units must establish a system for disseminating information to parents about the importance of rescreening.

D. Notification of Screening Results

1. Providers are notified immediately of positive screens by telephone. Otherwise, submitters should receive the result slip from the State Central Laboratory within two to three weeks. Submitters may call the Central Lab for results 10 days after submission. The telephone number for the Central Lab is 504-568-5371. Results are also available to submitters 24 hours a day, 365 days a year through the Voice Response System with FAX (VRS) which is accessed by using a touch tone telephone. Information on using VRS can be obtained by calling the Genetic Diseases Program Office at 1-800-871-9548. To assist the pediatrician's office in the retrieval of the results on the initial specimen of the

infant at the first medical visit, the phlebotomist or nurse collecting the initial specimen should tear off the blue carbon of the Lab-10 form and give this to the parent or guardian.

E. Unsatisfactory Specimens. The accuracy of a test depends on proper collection of the blood spot. Specimens of unsatisfactory quality for testing will be indicated on the result slip. Training on collecting adequate specimens can be arranged by calling the Genetics Nurse at telephone number (504) 568-5070.

F. Medical/Nutritional Management

1. In order for a patient with PKU or other rare inborn errors of metabolism to receive the special formulas for the treatment of these disorders from the state's Genetic Diseases Program and/or Special Supplemental Nutrition Program for Infants, Women, and Children (WIC), the following guidelines must be met:

a. The patient must be a resident of the State of Louisiana.

b. The patient must receive clinical and dietary management services through a metabolic center to include a medical evaluation at least once annually by a physician who is board certified in biochemical genetics or a medical geneticist physician with written documentation of a medical evaluation and continuing consultation with a physician board certified in biochemical genetics. A licensed registered dietitian must also be on staff and be readily available for both acute and chronic dietary needs of the patient. Children less than one year of age must be seen by the dietitian and medical geneticist at least twice a year. Children greater than one year of age must be seen at least once per year by the dietitian and medical geneticist.

c. The patient must provide necessary blood specimens for laboratory testing as requested by the treating physician meeting the above requirements. Laboratory test result values for phenylalanine and tyrosine must be submitted to the Genetics Program Office by the treating medical center within 15 working days after data reduction and interpretation.

d. The patient must include dietary records with the submission of each blood specimen.

e. All insurance forms relative to charges for special formula must be signed and submitted by the parent or appropriate family member.

f. The parent or guardian must inform the Genetics Program Office immediately of any changes in insurance coverage.

g. If a patient fails to comply with these requirements, he/she will not be able to receive metabolic formula, medications and medical services through the Office of Public Health.

G. Acceptable Newborn Screening Testing Methodologies and Procedures for Medical Providers not using the State Laboratory. Laboratories performing or intending to perform the state mandated newborn screening battery on specimens collected on Louisiana newborns must meet the conditions specified below pursuant to R.S. 40:1299.1.

1. The testing battery must include testing for phenylketonuria (PKU), congenital hypothyroidism, biotinidase deficiency, galactosemia and the following hemoglobinopathies: sickle cell disease, SC disease, thalassemias, E disease and C disease.

2. The laboratory must perform the newborn screening testing battery on at least 50,000 specimens a year unless the said laboratory has been routinely performing the full screening battery since January 1, 1995.

3. A laboratory must perform the complete battery at one site. Using two laboratories for completion of the total battery is unacceptable as this increases the risk of error and delay in reporting.

4. When using dried blood spots, only specimen forms using filter paper approved by the Centers for Disease Control (CDC) are acceptable.

5. Only the following testing methodologies are acceptable without prior approval.

Disease	Testing Methodology
PKU	Flourometric Tandem Mass Spectrometry Guthrie Bacterial Inhibition Assay Phenylalanine level cut-off: >3 mg/dL, call Genetics Office immediately for obtaining phenylalanine/tyrosine
Congenital Hypothyroidism	Radioimmunoassay (RIA) or Enzyme Immunoassay (EIA) Methods for T4 and/or Thyroid Stimulating Hormone (TSH) which have been calibrated for neonates
Biotinidase Deficiency	Qualitative or Quantitative Enzymatic Colorometric or Flourometric
Hemoglobinopathies (Sickle Cell)	Cellulose acetate/citrate agar Capillary isoelectric focusing (CIEF) Gel isoelectric focusing (IEF) High Pressure Liquid Chromotography (HPLC) DNA Analysis Sickle Dex – NOT acceptable Controls must include: F, A, S, C, E Result Reporting: by phenotype Positive/negative is NOT acceptable
Galactosemia	Galt enzyme assay Total Galactose
New Food and Drug Administration approved methodologies may be used if found to be acceptable by the Genetic Diseases Program. Approval should be requested in writing 60 days before the intended date of implementation (see Genetic Diseases Program mailing address below). Requests for approval will be based on documentation of FDA approval and an in-house validation study of said methodology.	

6. The laboratory must comply with the regulations for proficiency testing as mandated in the Clinical Laboratory Improvement Amendments of 1988 (CLIA 88 Section §493.1707). When using dried blood spots, the laboratory must participate in the proficiency testing program of the Centers for Disease Control. The laboratory must report all proficiency testing results to the Genetic Diseases Program Office within one month of receiving the report from the proficiency testing provider.

7. The laboratory must be able to provide test result data to physicians and nurses on their specific patients by telephone and by FAX or by use of the internet, 24 hours a day 365 days a year.

8. Mandatory Reporting of Positive Test Results Indicating Disease

a. To ensure appropriate and timely follow-up, positive results must be reported, along with patient demographic information as specified below to the Genetic Diseases Program Office either by FAX at (504) 568-7722 or by telephone at (504) 568-5070 and followed up by the mailing of the information to the following address: Genetic Diseases Program, P.O. Box 60630, Room 308, New Orleans, LA 70160-0630.

b. Specific time deadlines after data reduction and interpretation for reporting positive results indicating probable disease to the Genetics Office:

i. PKU: report a phenylalanine level of >3 mg/dL on the initial or repeat blood specimen within 2 hours;

ii. galactosemia: report test results on the initial or repeat blood specimen within 2 hours;

iii. congenital hypothyroidism: report confirmatory test results within 24 hours;

iv. biotinidase deficiency: report results within 24 hours;

v. sickle cell disease and other hemoglobinopathies: report results of FS, FSC, FSA, FSE, FS-other, FC, FCA and FC-other from initial specimens within 24 hours.

c. The specified information to be reported:

i. child's name;

ii. parent or guardian's name;

iii. child's street address;

iv. child's date of birth;

v. child's sex;

vi. child's race;

vii. parent's telephone number;

viii. collection date;

ix. test results;

x. primary care physician;

xi. age at collection (< or > 48 hours old);

xii. birth weight;

xiii. full term or premature or gestational age; and

xiv. transfusion:

Yes___ Date of last transfusion No___ (if available)

9. Provision of Follow-up Services. To ensure that reporting time deadlines are met for every positive result indicating probable disease under b above, a follow-up system must be in operation. The protocol for a follow-up system may rely on the submitting hospital for the follow-up action which must include the following.

a. Locate the infant and ensure diagnostic and medical care:

i. telephone call to medical provider within 24 hours of positive lab result;

ii. if there is no medical provider available, a telephone call should be made to parent/guardian;

iii. if the parent/guardian does not have a telephone, then notify them by certified and regular mail;

iv. if there is no response to mail within five days, a home visit should be made;

v. report to the Genetic Diseases Program Office all patients with suspect results who are unable to be located.

b. Results of repeat testing should be obtained.

i. If results are normal, the case can be closed.

ii. If results are abnormal, the case must be reported to the Genetic Diseases Program Office.

10. Reporting requirements of private laboratories to the Genetic Diseases Program Office for public health surveillance and quality assurance purposes.

a. The laboratory must submit quarterly statistical reports to the Genetic Diseases Program Office that indicate the number of specimens screened by method, the number of specimens unsatisfactory for testing, the number normal and positive, and for screening of hemoglobinopathies, the number by phenotype (see Genetics Office address in Paragraph G.7).

b. Effective July 1, 2001, the laboratory must also report to the Genetic Diseases Program Office via electronic transmission newborn screening results on all Louisiana newborns screened monthly or quarterly. The method of transmitting as well as the reporting must be by diskette or another mutually agreed upon form of electronic transmission. The file format and data layout will be determined by the Genetic Diseases Program. Essential patient data is the following:

i. child's first name;

ii. child's last name;

iii. mother's first name;

iv. mother's last name;

v. mother's maiden name (optional);

vi. child's street address;

vii. child's city;

viii. child's state;

ix. child's zip code;

x. child's parish (optional);

xi. child's date of birth (format: mm/dd/yyyy);

xii. child's sex;

xiii. child's race (format: (W)hite, (B)lack, Native America, Asian, other, Hispanic);

xiv. mother's social security number (format: 999-99-9999).

11. The laboratory must register by letter with the Genetic Diseases Program of the Office of Public Health

each year. This letter must contain the following and be received in the Genetic Diseases Program Office by February 1 each year:

a. assurance of compliance with the requirements described in Paragraphs 1-10 above;

b. the type of testing methodologies used;

c. the number of specimens projected to be tested or actually tested annually;

d. the type of specimen(s) used, i.e., filter paper or whole blood;

e. reporting format for positive/abnormal test results.

12. Guidelines and recommendations on quality assurance of newborn screening from nationally recognized committees and authors should be considered in the establishment and operation of a newborn screening system[2].

Reference

[1]American Academy of Pediatrics, Committee on Genetics: New Issues in Newborn Screening for Phenylketonuria and Congenital Hypothyroidism. *Pediatrics* 1982; 60-104-6.

[2]References pertaining to Subsection G:

a. Committee on Genetics, American Academy of Pediatrics Issues in Newborn Screening. *Pediatrics* 1992;89:345.

b. CORN Newborn Screening Committee, Council on Regional Networks for Genetic Services. U.S. Newborn Screening System Guidelines: Statement of the Council of Regional Networks for Genetics Services. Screening, 1 (1992 pp. 135-147).

c. Andrews L *Legal Liability and Quality Assurance in Newborn Screening.* Chicago, American Bar Foundation (1985), pp. 82-83.

d. National Committee for Clinical Laboratory Standards (NCLS) Standards for Blood Collection on Filter Paper for Neonatal Screening. Document LA4-A2 July 1992.

e. Committee on Assessing Genetic Risks, Division of Health Sciences Policy, Institute of Medicine *Assessing Genetic Risks* National Academy Press, Washington, D.C. (1994).

f. Clinical Laboratory Improvement Amendments, 1988. Health Care Financing Authority (HCFA).

AUTHORITY NOTE: Promulgated in accordance with R.S. 40:5 and R.S. 40:1299 et seq.

HISTORICAL NOTE: Promulgated by the Department of Health and Human Resources, Office of Preventive and Public Health Services, LR 13:246 (April 1987), amended by the Department of Health and Hospitals, Office of Public Health, LR 17:378 (April 1991), LR 18:1130 (October 1992), LR 20:1386 (December 1994), LR 33:301 (March 1997), LR 27:545 (April 2001), LR 29:1490 (August 2003).